American Journal of Medical Genetics 66:247–249 (1996)

Genetic Drift

The Christmas Present

Robert W. Marion

The Center for Congenital Disorders, Department of Pediatrics, Montefiore Medical Center/Albert Einstein College of Medicine, Bronx, New York

Telling parents that their child is affected with a genetic disease is undoubtedly the worst part of our job. Often painful and always distressing, it's unfortunately something we cannot avoid: so many of the conditions with which we clinical geneticists deal have such terrible prognoses. In these situations, we are forced to act as both judge and jury. Because he is affected with a disorder, we have found the child guilty, and it's our role to condemn him to death and his family to a life of grief. And, in most cases, once the diagnosis has been confirmed and the news delivered, we're helpless to do anything to avert the predetermined outcome. We can't fix the child, and we often can't significantly alter the course of his illness. At best, in an attempt to make his existence acceptable, all we can really hope to do is orchestrate some of the events of the child's life.

That's what happened the first time I saw the Sweeneys.

It was the Friday before Christmas, and the Garwood Children's Rehabilitation Center had that feeling of forced festivity that tends to envelop children's hospitals that time of year. It seemed as if every last inch of the place was decorated with tinsel and blinking lights, garishly-dressed evergreens, and giant menorahs. In spite of all these carefully-arranged, cheerful decorations, a feeling of helplessness and hopelessness still hung in the air.

Amy McDonald, a second year fellow in genetics, and I were standing in the hallway talking with Benjamin Sontag, the Executive Director of Garwood, when we first saw the Sweeneys. Mother, father, and son were rapidly walking down the hall toward the outpatient department. From my position, I could only see the boy in profile; Amy was able to see him straight on. We saw the child for only a few seconds as, clutching the hands of his parents for support, he walked past us. But that brief glimpse of the boy was enough. His large head, his coarse facial features, his thick, claw-like hands, and his stiff, spastic gait, told us more than either the fellow or I needed to know. I looked at Amy, she looked at me, and our jaws dropped.

"What's wrong?" Ben asked, noticing the change in our demeanor.

"That boy . . ." Amy began.

"What about him?" Ben asked, looking down the hall, probably taking notice of this family for the first time.

"He's got a mucopolysaccharidosis," I replied.

"I'm sure he does," Ben said. "What's a mucopolysaccharidosis?"

"A storage disease," Amy responded. "They're a group of disorders that usually cause degeneration of the central nervous system and early death."

"You can tell all that just by seeing the boy pass in the hall for a few seconds?" Ben asked.

"Ben, if we couldn't tell all that just by seeing him pass in the hall, we wouldn't deserve to be your genetics consultants," I replied.

"It looks to me like he probably has Hunter syndrome." Amy added.

"Hunter syndrome I know about," Ben said. "At least I know enough to know that Hunter syndrome is not a good thing to have."

Amy and I nodded. "We better go find out who that kid is and why he's here," I said.

By this point, the family had disappeared around the corner that led to the outpatient department. Amy and I followed down the hall after them and, seeing that the parents had settled themselves in the waiting area, and that the boy had quickly begun to attack the waiting area's well-stocked toy chest, we entered the reception station.

"Who's that kid?" I asked Joanne, the clerk.

"Thomas Sweeney," Joanne replied, "your first patient of the afternoon."

After pulling the boy's chart from the "To Be Seen" basket, I began riffling through it. I found the genetics consult request form and as I read it, my heart sank. "He's a three-year-old who was referred to us for evaluation by Eileen Woods, the audiologist," I mumbled in Amy's direction. "He has hearing loss and global developmental delay."

"Does it say anywhere that he has Hunter syndrome?" Amy asked.

Received for publication February 26, 1996; revision received April 8, 1996.

Address reprint requests to Robert W. Marion, Department of Pediatrics, Montefiore Medical Center, 111 East 210 St., Bronx, NY 10467.

Essays in "Genetic Drift" emphasize the human side of genetics. Where appropriate, names and locations are changed to maintain confidentiality.

I turned the pages of the chart. Results of an audiogram revealed that the boy had a mild-to-moderate conductive hearing loss. A note from Eileen briefly outlined the boy's history: a year before, the note reported, he had been able to say a half-dozen words; more recently, not only had he not gained any new words or started putting them together in sentences, he'd apparently lost all ability to communicate verbally. Except for these notes and some insurance information, the chart was empty. "No, no diagnosis listed," I said to Amy. "We'd better go talk to Eileen."

We found the audiologist in her office, doing some paperwork. "Thomas Sweeney's here to see us," I said, taking a seat across from Eileen's desk. "Just looking at him in the hall, we're pretty convinced he has either Hunter syndrome or some other mucopolysaccharidosis. Eileen, do you know if anyone's raised this possibility with the parents in the past?"

The audiologist smiled sadly and shook her head. "Apparently not," she replied. "I saw him for the first time about two weeks ago. His parents made the appointment themselves because Tommy's speech is so far behind. They've been worried about him for over a year, but their pediatrician has been blowing them off, telling them that he's just a little slow in getting started but that he'll eventually catch up."

"Terrific," Amy interrupted. "That's very helpful."

"The parents knew Tommy was more than just a little slow," Eileen continued, "and since they weren't getting any help from their doctor, they finally decided to take matters into their own hands. When I saw him, I knew he had something, but I wasn't sure what it was."

"How did you get them to make an appointment to see us?" I asked.

"That was easy. Since the audiogram showed that he had significant hearing loss, I told the parents that sometimes these problems were inherited, and that it might be a good idea for Tommy to see a geneticist. It didn't take a lot to convince them; they're so concerned, they made the earliest possible appointment with you. The mother told me that they've put off having more children until they get an answer about what's causing Tommy's problem. They're very nice people, and they're scared to death."

"They have good reason to be," I said. "You remember what happens to kids with Hunter syndrome?"

Eileen nodded her head. An excellent audiologist who'd worked at Garwood for a long time, Eileen Woods had had a lot of experience with children and adolescents with various forms of mucopolysaccharidoses. Over the years, she'd carefully and methodically documented the inevitable slow, steady deterioration of their speech and hearing.

"Do you have any feel for how they're going to take this news?" Amy asked.

The audiologist hesitated for only a few seconds. "They're going to be devastated," she said.

I nodded my head, and Amy and I left her office.

* * *

Because it was the Friday of a holiday weekend, most everyone at Garwood had planned to leave work early, in an attempt to get a jump on the traffic. Before starting with the Sweeneys, I wanted to make sure that all the tests needed to confirm the suspected diagnosis could be performed. Amy and I went into our office and started making phone calls. I called the X-ray tech, to see if she'd be able to do a skeletal survey in about an hour, while Amy called the lab, to make sure they'd be open to draw the blood, collect the urine, and box all the specimens so that they could be mailed to our reference lab. In neither case were the people on the other end of the phone exactly overjoyed about the prospects of working up a child for a mucopolysaccharidosis that afternoon. But both reluctantly agreed that if we got the patient to them by three o'clock, they'd do what needed to be done.

After hanging up the phone, I sat back in my desk chair and sighed. "I hate this part," I said sadly to Amy. "I hate having to tell them. But we've got to do it, so we might as well get it over with."

As I was beginning to rise, Amy said, "Bob, are you sure you want to do this?"

"What do you mean?" I asked, and sat back down in my chair.

"Look, it's three days before Christmas. If this boy has Hunter syndrome, he's had it for three years, right?"

"Right," I replied.

"And if he's had it for three years, isn't he also likely to still have it next week and the week after that?"

I nodded my head.

"Right," Amy continued. "Christmas is going to be hard enough for these people as it is. Look at what they're going through: they know something's wrong with their only child, something that's preventing him from being able to speak. It's got to be frustrating for them, but at least without a name for the condition or a prognosis, they still can hold onto some hope. I'm sure that, deep down, they both believe that whatever's wrong can be fixed with either medication or surgery, or that it might even resolve on its own. Do you agree?"

I again nodded my head, and Amy continued, "Now think of what Christmas will be like if we tell them that their son has an incurable neurodegenerative disease that will not only prevent him from ever being able to communicate, but will also wind up killing him by the time he's twenty."

Thinking about Amy's words for only a few seconds, I nodded my head. "You're absolutely right, Amy," I finally responded. "Telling them today would destroy whatever joy they might have had over the next week. And there's nothing that would be gained from telling them. No, you're right. There's absolutely no reason for us to tell these people today that we think their son has Hunter syndrome."

"It would be different if we had some treatment to offer, or if there was a pregnancy involved," Amy went on. "But there is no magic pill, and Eileen just told us that they're holding off having more children until they know for sure exactly what's wrong with the boy. Making the diagnosis today, next week, or even next month won't change anything."

I continued to nod my head. "There's only one problem," I said. "They're already here for their appoint-

ment. I don't feel comfortable seeing them and not telling them what we think."

The office remained silent as both Amy and I thought through this problem, trying to come up with a solution. Finally, reaching for the phone, I broke the silence. "I'm just going to have to lie," I said. As I dialed the number of the outpatient department's reception station, I added, "Now, Amy, I want you to understand, I'm not encouraging this kind of behavior. But occasionally, not telling the truth is in the best interest of the patient and his family."

After two rings, the clerk picked up the phone. "Joanne," I said, trying to sound as pained as possible, "this is Bob Marion. I'm sorry to do this. I know I have a patient waiting out there, but I've developed a terrible migraine headache. I have to take some medication and lie down for a while. Would you apologize to them for me and reschedule them for the first Friday in January?"

* * *

When I emerged from my office a half hour later, miraculously free of my headache and ready to see my next patient, the Sweeneys were gone. Amy and I saw the remainder of the patients scheduled for the genetics clinic that afternoon. When we left a nearly deserted Garwood, I was still thinking about the Sweeneys, still turning over in my mind whether we'd truly done the right thing.

During their visit in early January, Amy and I finally had to face telling them of our concerns. As expected, the session was a rough one. The couple, understanding all too well what we were saying, at first denied that Tommy had anything worse than just some mild hearing loss, but ultimately, faced with all the evidence we presented, they came to accept our conclusion. As they held onto each other, both crying softly, we told them that no matter what the workup showed, regardless of whether Tommy had Hunter syndrome or not, we'd be there for them, always available to offer information and advice, to follow their son during the years to come, and just to talk.

We did a complete evaluation of Tommy that afternoon. X-rays of the boy's bones showed that he had dysostosis multiplex; a urine sample showed equal but markedly elevated excretion of dermatan sulfate and heparan sulfate; and an assay of iduronate sulfatase performed on a blood sample revealed a complete absence of enzyme activity, confirming the diagnosis of Hunter syndrome.

It's been more than a month now since I called Mr. Sweeney's office to tell him that the lab tests had confirmed what we all already knew. The boy, who almost immediately began a course of vigorous physical, occupational, and speech therapy, has already shown some improvement in his motor skills, but his lack of speech continues unchanged. I check in with Tommy's mother and father at least once a week, trying to assure myself that they're weathering this emotional storm. They seem to be doing as well as can be expected. Mrs. Sweeney told me last week that although falling asleep at night continues to be difficult, she and her husband have finally achieved something of a milestone: they've been able to make it through the day without crying.

While at Garwood for clinic yesterday, I spoke with Mr. Sweeney. We were talking about how life had changed for the family since Tommy's diagnosis was confirmed, and, for the first time, we discussed Christmas. "You can't imagine how perfect that day was," he said. "Our house was filled with laughter and happiness. Our parents came for dinner, we exchanged presents, and of course, as the only grandchild on either side, Tommy was the center of attention. When I think of what's happened since then, I can't believe how happy we all once were. I'll always remember that last Christmas; it was the last time that our world seemed anything like normal."

After a few minutes, we said good-bye and after hanging up the phone, I just sat in my office chair, thinking. I've turned over in my mind many times since that late December day whether or not telling that lie was the right thing to do. Though, in general, it's difficult to justify lying to patients, I've decided that in some situations, in some circumstances, it may be acceptable. Holding back the news from the Sweeneys on that Friday before Christmas was one of those justifiable situations; by not seeing the family, Amy and I made sure that they'd have one last memorable holiday before their lives were irretrievably changed. That lie was our Christmas present to the Sweeneys.